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☐ 1: Gene 2001 Jul 25;273

(1):89-96

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ELSEVIER SCIENCE **FULL-TEXT ARTICLE**

Two new genes from the human ATP-binding cassette transporter superfamily, ABCC11 and ABCC12, tandemly duplicated on chromosome 16q12.

Tammur J, Prades C, Arnould I, Rzhetsky A, Hutchinson A, Adachi M, Schuetz JD, Swoboda KJ, Ptacek LJ, Rosier M, Dean M, Allikmets R.

Department of Biotechnology, Institute of Molecular and Cell Biology, Tartu University, Tartu, Estonia.

Several years ago, we initiated a long-term project of cloning new human ATP-binding cassette (ABC) transporters and linking them to various disease phenotypes. As one of the results of this project, we present two new members of the human ABCC subfamily, ABCC11 and ABCC12. These two new human ABC transporters were fully characterized and mapped to the human chromosome 16q12. With the addition of these two genes, the complete human ABCC subfamily has 12 identified members (ABCC1-12), nine from the multidrug resistance-like subgroup, two from the sulfonylurea receptor subgroup, and the CFTR gene. Phylogenetic analysis determined that ABCC11 and ABCC12 are derived by duplication, and are most closely related to the ABCC5 gene. Genetic variation in some ABCC subfamily members is associated with human inherited diseases, including cystic fibrosis (CFTR/ABCC7), Dubin-Johnson syndrome (ABCC2), pseudoxanthoma elasticum (ABCC6) and familial persistent hyperinsulinemic hypoglycemia of infancy (ABCC8). Since ABCC11 and ABCC12 were mapped to a region harboring gene(s) for paroxysmal kinesigenic choreoathetosis, the two genes represent positional candidates for this disorder.

PMID: 11483364 [PubMed - indexed for MEDLINE]

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